ABSTRACT

Title: Methods

This invention relates to polymorphisms in the human pyruvate dehydrogenase E1β (PDH E1β) gene. The invention also relates to methods and materials for analysing allelic variation in the PDH E1β gene, and to the use of PDH E1β polymorphism in the diagnosis and treatment of diseases in which modulation of pyruvate dehydrogenase activity could be of therapeutic benefit, such as diabetes, asthma, obesity, sepsis and peripheral vascular disease. In particular, the invention is based on the discovery of a single nucleotide polymorphism in the coding region of the human PDH E1β gene, and three single nucleotide polymorphisms in the 3' untranslated region (3'UTR) of the human PDH E1β gene.